

Bardet-Biedl Syndrome GeneReview: Molecular Genetics

Table 7. MKKS/BBS6 Normal Allelic Variants

Gene	Sequence Change	Exon	Coding Frequency	References
<i>BBS6/MKKS</i>	c.-675_674ins [GTGGCGGCCT]	1A	2.25%	Slavotinek et al 2002
<i>BBS6/MKKS</i>	c.-74G>A	3	0/94	Slavotinek et al 2002
<i>BBS6/MKKS</i>	c.117C>T; nil	3	Not done	Slavotinek et al 2002
<i>BBS6</i>	p.G49V	3	8.3%	Stone et al 2000
<i>BBS6</i>	c.534C>T; nil	3	Not done	Slavotinek et al 2002
<i>BBS6</i>	p.R517C	6	6.3%	Stone et al 2000
<i>BBS6</i>	p.G532V	6	6.4%	Stone et al 2000